Two Novel Mutations in Gene *SPG4* in Patients with Autosomal Dominant Spastic Paraplegia

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Abstract—Hereditary spastik paraplegias (HSP) are a group of neurodegenerative disorders with primary lesion of the pyramidal tract. The most frequent autosomal dominant form of the disease in Europeans is HSP associated with mutations in the spastin gene (*SPG4*). Analysis of the gene *SPG4* was carried out in 52 unrelated families with HSP from Bashkortostan by SSCP and following sequencing. Previously undescribed frameshift mutations c.322del29 (p.Val108SerfsX18) and c.885del10 (p.Thr295ThrfsX16) were detected in two unrelated families. Clinical studies have shown that, in both families, the disease corresponds to an uncomplicated form of hereditary spastic paraplegia, a main feature of which is the lower spastic paraparesis without any other symptoms.

Keywords: hereditary spastic paraplegia, gene *SPG4*, mutations, Republic of Bashkortostan

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